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ANATOMICAL STUDY OF A SHORT-LIMBED DWARF, WITH
SPECIAL REFERENCE TO OSTEOGENESIS IMPERFECTA
AND CHONDRODYSTROPHIA FŒTALIS.¹

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IN the winter of 1901 there fell into my hands the body of one Thomas Wilson, concerning whom the following facts are all that are known at this time: He could not walk very well and supported himself by selling lead-pencils and chewing-gum from a roller chair. He was a hard drinker. Two persons who knew him during life state that in their opinion he was of ordinary intelligence. It is unfortunate that nothing is known of his early life. He died January 5, 1901, in a so-called "bum lodging-house," at the supposed age of forty-five years. A post-mortem examination was made by the physician to the coroner's office, and rupture of the gall-bladder is mentioned in the certificate as the chief cause of death. The body was then kept in a refrigerator until March 7, 1901, when a second post-mortem was made, with the following result:

There is a sutured post-mortem incision from the top of the sternum to the pubes, and there is also a sutured incision in the left arm. The body is diminutive in size; the head and trunk are more nearly normal in size; the lower extremities are perceptibly shorter in proportion. In the lower extremities, especially the legs, the skin seems to be looser than usual; there is a moderate curvature of the spine in the thoracic region, the convexity extending forward and to the left, with a marked curve in a forward direction in the lumbar region. The bones of the extremities are the only ones that show any abnormalities on external examination. The long bones are the seat of various curvatures and nodular enlargements, especially at the articular ends. In the left humerus there is a distinct interruption of the continuity in the lower third, but this has not the appearance of a recent fracture, and crepita-

¹ The skull is described by Charles A. Parker, M.D., and the thyroid examined chemically by H. G. Wells, M.D.

tion is not obtained. (In the X-ray photographs of the body, kindly made by Dr. Smith, resident physician of the Presbyterian Hospital, the bones of the skeleton are seen to be excessively porotic, anomalous in form, the long bones being greatly swollen at the ends.) The right knee-joint is larger than the left, and there is a swelling in the head of the right tibia, which crackles on pressure. Incision into the joint shows a soft mass from which red hemorrhagic material can be scooped out. There are no evidences of decomposition, but there is considerable drying up of the tips of the fingers and toes, and of the nose, lips, and penis; the latter seems to be very small. The nose appears rather thin and sharp, and the root does not seem to be any more deeply inserted than is ordinarily the case. There is no distinct depression at the root. Two small testicles are felt in the scrotum. Otherwise the external examination reveals no further morbid lesions.

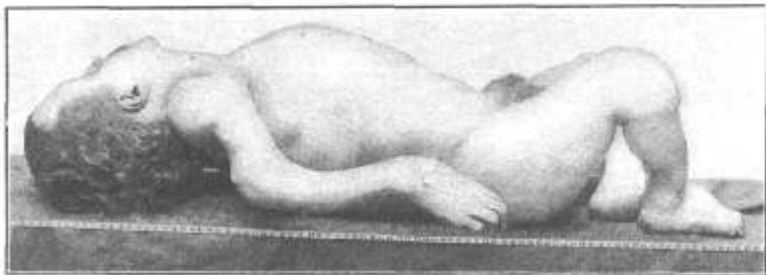
The following measurements were made:

Length of body	95 cm.
Estimated weight	75 lbs.
Length of right upper extremity	48 cm.
" left "	49 "
" hands	15 "
Breadth of hands	6 "
Circumference of middle of right arm	30 "
" " left "	30 "
Length of right lower extremity	37 "
" left "	36 "
" feet	13 "
Breadth of "	7 "
Vertical diameter of patella	3 "
Circumference of right knee	28 "
" of left knee	24 "
" of middle of right thigh	27 "
" of middle of left thigh	25 "
" of head	30 "
Distance from tip of chin to highest point of head	27 "
" " root of nose to chin	11 "
" " glabella to occipital protuberance	21 "
" " to protuberance over top of head	35 "
" between zygomatic arches	12 "
" from meatus to meatus over top of head	36 "
" between malars	11 "
" " angles of lower jaw	8.5 "
" from tip of chin to pubes	42 "
Length of spine (sacrum 7, lumbar 12, thoracic 18, cervical 11)	48 "
Circumference of neck	26 "
" at shoulders	69 "
" of chest at nipples	69 "
" of abdomen at navel	57 "
" of pelvis at level of superior spine	57 "

The body cavities are found filled with sawdust soaked in formalin. The heart, lungs, spleen, pancreas, liver, kidneys and lower urinary organs, testicles, and organs of the neck do not present any gross lesions recognizable at this time. All the organs are smaller than those of a normally developed, full-grown man. The heart weighs 145 grammes; the kidneys, 105 grammes; the spleen, 30 grammes; and the thyroid, which is symmetrical and flattened, 9 grammes.

The diameter of the aorta at its beginning is 9 millimetres; in the middle of the descending portion it is 1 centimetre.

FIG. 1.



Photograph of the body. Note relative shortness of the limbs.

The gall-bladder does not show any changes, but there is a tear across the centre of the free aspect.

The bones of the skull are rather thin and cut easily with the saw. The brain is quite soft, and the hypophysis weighs 0.65 grammes.

The skeleton was prepared for articulation and mounting.

HISTOLOGICAL EXAMINATION.

Heart. There are no special changes. The number of nuclei in the muscle cells seems rather large. The striations are absent, the fibres are narrow, and branch and anastomose unusually freely.

Thyroid. In some parts of the gland there is no evidence of thyroid structure present—the gland here appears to have been entirely replaced by fibrous and muscular tissue. For the most part the typical follicular structure is replaced by fibrous tissue with oblong and variously-shaped spaces filled with rather small cells of various shapes, the nucleus staining diffusely, the cell body being without any distinct wall (Fig. 3), the nuclei are relatively large, the surrounding finely granular cytoplasm staining a light pink with eosin. In no place do these cells assume a distinct cuboidal form, and, as stated, there is no trace of any follicular arrangement. Small masses of homogeneous or finely granular colloid mate-

FIG. 2.

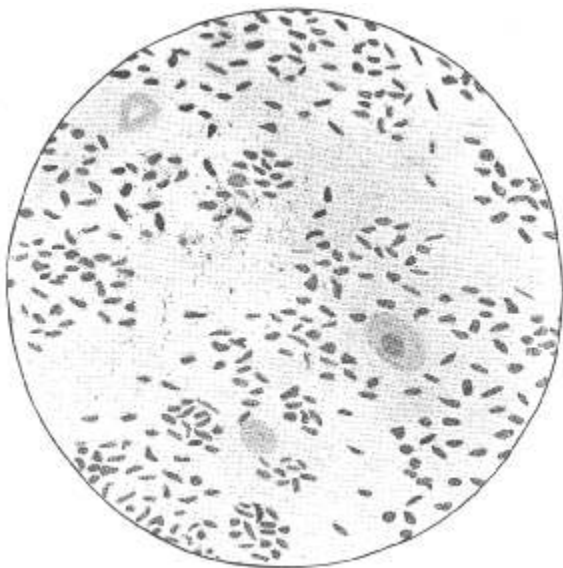


Photograph of the body.

rial occur here and there. Generally but one or two such masses appear in the field of a No. 3 objective. Sometimes colloid masses lie in spaces containing but few cells, the walls being formed by fibrous tissue. At other times the spaces are lined with endothelial cells. The thyroid contains a moderate number of bloodvessels of normal appearance; in some of the larger arteries at the margins there is fibrous proliferation in the intima and calcification of the elastica.

Hypophysis. The structure seems quite normal. There are small masses of colloid in a few of the follicles. There are no distinctly eosinophilic or cyanophilic cells present, the cell bodies generally assuming a light pink color in hæmatoxylin and eosin sections.

FIG. 3



Fibrosis and atrophy of the thyroid, with occasional colloid remnants.

Bones. Sections of bony tissue from various bones, especially the upper end of the right tibia, show an exceedingly spongy structure; for the most part only isolated fragments of bone occur, hardly any complete Haversian systems being present; there is no evidence of excessive lacunar absorption or halisteresis, but the number of bone corpuscles seems larger than usual. The marrow is fatty.

CHEMICAL EXAMINATION OF THE THYROID (by Dr. H. G. Wells). The thyroid gland, except such part as had been used for histological examination, was received in alcohol. In order to avoid possible loss from solution in the alcohol the fluid was evaporated to dryness in the incubator and the residue added to the gland substance, which was similarly dried. The weight of the substance combined was 0.766 grammes. Analysis was performed in the same manner as in the series previously studied, that is, by the modified Baumann's method. The residue from the infusion, after being evaporated, was shaken with 10 c.c. of chloroform, and no coloration whatever was imparted

to it. Therefore, if any iodine at all was present it was but a minute quantity, not over 0.05 milligramme. The average amount of iodine per gramme of dried thyroid in residents of Chicago has been found to be about 2 milligrammes.¹

FIG. 4.



Photograph of skeleton. Relative length of upper extremities in this and in the two succeeding figures due to imperfect articulations, especially at the shoulder-joints.

EXAMINATION OF THE MOUNTED SKELETON (Figs. 4, 5, and 6). The skeleton as a whole is small and light, the total weight, including

¹ Concerning the method of analysis, see H. G. Wells, "The Physiology and Therapeutics of the Thyroid Gland and its Congeners." *Journal of the American Medical Association*, October 30, and November 6 and 13, 1897.

an iron rod to hold the spine together and copper wires at the majority of the joints, being only 1817 grammes. The bones in general are light and spongy; there is no sclerosis anywhere, the external as well as the internal architecture presenting many and striking variations

FIG. 5.



Photograph of skeleton. Left lateral view.

FIG. 6.



Photograph of skeleton. Right lateral view.

from the normal. The usual normal anatomical markings are greatly disturbed, often absent or not recognizable. In the following description only the essential and most remarkable departures from the normal type are mentioned briefly. The spine contains the usual

number of vertebræ. There is a slight curvature forward and to the left in the upper thoracic region, resulting in some elevation of the left shoulder. At the junction between the eleventh and twelfth thoracic vertebræ occurs a sharp bend forward almost to a right angle. The bodies of the twelfth thoracic and of the first lumbar vertebræ are much flattened, being 1 cm. in thickness in front. At the lumbosacral junction the sacrum bends backward nearly to right angles, but the lower end of the sacrum is bent forward at an acute angle, the coccyx projecting between the ischia.

The shoulder-blades are small and thin, and of a distinct foetal character. In the right subscapular fossa is a defect 2 cm. in diameter, with smooth but thin margins. There are no distinct glenoid cavities, the enlarged heads of the humeri being partially received into a concavity formed by the elongated coracoid processes, which make a semicircle, and parts of the margins of which seem to form articular surfaces.

The normal curves of the clavicles are exaggerated. The right clavicle is 10 cm. long, the left 11.5 cm.

There is a slight thickening at the costo-chondral junctions. The heads and necks of the first five right ribs are more or less absorbed and amalgamated with the corresponding vertebræ by osseous tissue. The third, fourth, and fifth are broken at or near the angles; in the case of the third and fourth there seems to be false joints at the sites of the fractures; in the case of the fifth the fracture may be post-mortem. The vertebral ends of the first and second ribs are embedded in a rather irregular bony growth. There is an old ununited fracture, with smooth surfaces in the sixth right rib, about 6 cm. from the costo-transverse junction; another in the sixth left rib, about 3 cm. from the spine.

The humeri are misshapen, enlarged at the ends, and in the case of the right humerus, which is bent almost to a semicircle, the shaft is quite thick. The heads of the humeri are swollen into irregularly globular masses, 4.5 cm. in diameter, and composed of crumbling, spongy bone. At the junction of the lower and middle third there is a break in the left humerus; the ends are somewhat swollen and irregular; the opposing surfaces are fairly smooth; for some distance on each side the bone is thin and slender. The right humerus is 17 cm. long, the left 16 cm.

The bones of the forearm are more or less curved, especially those of the right forearm, but there is no marked enlargement of the articular ends. At the junction of the middle and lower third of the right radius is a fusiform swelling such as might be caused by the healing of fracture. The right ulna is 17.5 cm. long; the left 19 cm.; the right radius 17 cm.; the left 19.5 cm.

The bones of the carpi, the metacarpal bones, and the phalanges seem normal in shape and size.

The pelvis presents marked deformities (see Figs. 4, 5, and 6). The bones are thin, light, and fragile. The alæ, especially in the regions of the fossæ iliacæ, are quite like parchment, and there are two oval defects with thin but smooth margins in the left and one in the right, from 1 cm. to 1.5 cm. in diameter. At each acetabulum the bone seems to have been pushed toward the sacro-iliac articulation, so that the distance between the corpus ossis-ischii and the sacrum on the left side is less than 1 cm., and on the right side about 2 cm. There is no

distinct acetabular rim. The superior-posterior parts of the acetabular rims have large facets for articulation, with reciprocal surfaces on the anterior portion of the superior surfaces of the necks of the femurs. The foramina obturata face each other, the incisuræ acetabuli being very large, 3.5 cm. and 4 cm. in diameter. The pelvis is of an irregular hour-glass form. Diameters: At inlet—transverse, 10 cm.; antero-posterior, 4 cm.; oblique, 8 cm. Of cavity—transverse, 2.5 cm.; antero-posterior, 9 cm. At outlet—transverse, 6 cm.; antero-posterior, 6 cm.

The femurs are similar in general outline, both describing a large curve with the convexities toward each other. The lower ends are greatly expanded and contain large spaces; the lower end of the right femur has crumbled away considerably. The neck is short, relatively thick, the head large, exceedingly spongy. The right femur is 19 cm. long, the left 20.5 cm.

In the dry condition the left tibia and fibula measure 17 cm. in length. The ends are swollen, especially the upper end of the tibia, which is expanded more or less mushroom-like. Both bones are curved, the convexity being directed in the front. The upper ends of the right tibia and fibula have crumbled away. These bones are also curved extensively, and 3 cm. above the ankle the fibula is united to the tibia by a flattened bridge 1 cm. wide.

The bones of the feet are exceedingly light, consisting of a very thin compact layer, externally and interiorly, of larger and smaller cavities separated by walls of extreme thickness. Many of the smaller bones crumbled down, and it was not possible to articulate the feet.

THE SKULL (Dr. Charles A. Parker). The skull¹ presents on lateral view an unusually high cranium overtowering a small receding face and protruding lower jaw. In front it has a narrow oval outline with high orbits and irregular teeth.

The maximum length is 17.2 cm., the maximum breadth 13.2 cm., with a corresponding circumference of 49.5 cm. The height from the basion, or anterior margin of the foramen magnum to the bregma, inside measurement, is 15 cm. Although a comparison of these figures with those averaged from ten normal skulls shows both the breadth and length to be slightly over 1 cm. short of the average, yet the height, which is increased by 2 cm. (the compensation relation), completely compensates for the circumferential contraction, as shown by the capacity of 1450 c.c., that of the average normal male skull.

The cephalic index is 76.7, hence the skull is of the meso-cephalic type.

The internal surface of the base presents little variation from the average skull in form or measurements; the foramen magnum is normal in size, shape, and location, as are also neighboring parts of the occipital bone, except the upper part of the tabular portion, which is composed of Wormian bones.

The thickness varies from 2 mm. laterally, in the temporal region, to 9 mm., in the frontal and occipital the average being 4 to 5 mm. The distance from the basion to the foramen cæcum is 1 cm. above the average, which, considering the shortness of the skull and normal

¹ Further consideration of the skull will be given by Dr. Parker in a separate article concerning Wormian bones.

location of the basion, appears disproportionate, but is explained by the absence of the frontal sinus and crest. The foramen is situated 1 to 2 cm. nearer the anterior surface of the skull and correspondingly farther from the basion. It is not unusual to find the distance from the glabella to the foramen over 2 cm., while in this skull it is less than 1 cm.

The distance from the basion to the occipito-sphenoidal synchondrosis is normal, as is also the distance from the latter to the anterior superior angle of the sphenoid at the cribriform suture. The cerebral surface of the presphenoid measures 2.5 cm., while the postsphenoid measures but 2 cm., giving an index of 125 obtained by dividing 100 times the former by the latter, the angle being very close to 100 or both parts equal. In a series of skulls examined the maximum was 113 and the minimum 87.

FIG. 7.



Photograph showing Wormian bones of cranial vault.

While the total length of the cerebral surface of the sphenoid is not affected, the disproportion between the two portions is unusual and constitutes the only noteworthy variation from the ordinary measurements of this region. Although somewhat asymmetrical in general outline, the entire cranium presents but slight variations from the normal measurements, the moderate circumferential constriction being accompanied by vertical elongation, with undiminished capacity, and the disproportion between the presphenoid and postsphenoid making no difference in the length of the floor.

The peculiar shape of the skull, with the recession of the face, may be represented by superimposing an outline of a median section of the skull upon a similar outline of an average normal skull, so that a line from the basion to the spheno-cribriform junction, the basal line of Huxley, is one exactly corresponding with a similar line in the other, their uppermost ends being at the same point (see Figs. 4 and 7).

Wormian Bones. The most striking feature of the skull is the extraordinary development and distribution of Wormian bones, numbering 172 (Figs. 7 and 8). They are most numerous in the posterior and lateral regions of the skull, supplanting the parietals, the squamous portions of the temporals, and the upper half of the tabular portion of the occipital. They are generally stellate in form, their numerous interlacing processes presenting an extensive pattern of delicate inlaid work. Several occur in the vertical and orbital portions of the frontal bones.

The inner surface is smooth, showing only the larger dentations, the finer process being limited principally to the outer layer. The parts free from them are the lower portion of the tabular plate, the condylar and basilar portions of the occipital, the mastoid and petrous portions of the temporals, the sphenoid and ethmoid bones, or those bones primarily developed in cartilage, the chondro-cranium.

FIG. 8.



Photograph showing Wormian bones of cranial vault.]

The sagittal suture is distinct throughout, and extends from the nasion to the chondro skeleton of the squamo-occipitalis, separating the parietal bones and dividing the frontal and membranous skeleton of the squamo-occipitalis into lateral halves. The coronal suture is well marked and sharply limits the forward extension of the Wormian bones, though one in the vertical portion of the right frontal bone and some in its orbital plate are in front of it. The lambdoid and squamous sutures cannot be traced. The sutures at the base show no special variations.

The frontal bone, which is divided into two parts by the persistent sagittal suture (metopic), presents a very high vertical portion and small orbital plates. In the upper part of the right half, bordering on the coronal and sagittal sutures, is a large quadrilateral Wormian

bone measuring 5 cm. by 9 cm., separated from the rest of the bone by a horizontal suture. Twelve small Wormian bones are located in the suture between the orbital plates and the *alæ parvæ* of the sphenoid, the cribriform plate and lateral masses of the ethmoid, the lacrymal, and the nasal processes of the superior maxillary bones.

Three-fourths of each parietal bone are represented by various forms of Wormian bones, varying in diameter from 3 mm. to 3 cm., while the remaining fourth consists of a single quadrilateral segment measuring 4 cm. by 8 cm., occupying the anterior superior angle. Bordering the coronal suture on either side below the larger segments are three or four of smaller size, ranging from 2 cm. to 3 cm. in diameter. The temporal ridges are indistinct or absent. On the inner surface the grooves for the meningeal arteries are well marked. The average thickness is 3 mm. to 4 mm.

The squamous portions of the temporals, together with the lower borders of the parietals, from which they are indistinguishable, present the most delicate tracing, in many bones the long interlacing processes being limited entirely to the outer layer.

The upper part of the squamo-occipitalis is formed entirely of Wormian bones.

The distribution by numbers in the different regions may approximately be given as follows: frontal bone, 12; parietals, each 40; temporals, each 25; occipital, 30.

Although so much of the cranium is composed of Wormian bones, yet the cranial anlage are well preserved, the principal sutures and bony areas being present and in their proper relations, showing that whatever general influences affected the skeleton, they were, in the skull at any rate, limited to osteogenesis and not of earlier origin or farther reaching in their effects disturbing the fundamental elements and interfering with the type of development as is observed in the perverted chondrogenesis of chondrodystrophia foetalis and frequently seen in monsters.

The same may be said of the rest of the skull; all the bones are present with their usual relations, though the imperfect development of some produces considerable deformity, most marked in the recession of the face.

Recession of the Face. When considered with the fact that the second and third molars, and the left lateral incisors in the upper jaw, and the left third molar and lateral incisor in the lower jaw have never erupted, it is evident that the marked recession of the face, reaching its maximum at the teeth, is due to perversions in the evolution of the latter, with consequent pernicious effects on the development of the jaws, which, together with the bones resting upon them, give the face its outlines.

The great disproportion between the height and width of the posterior nares, the latter exceeding the former by fully 1 cm., gives a broadening effect which, with the incomplete dentition already noticed, stamps the face anatomically of the infantile type, the general deformity being particularly conspicuous because of the adult cranium.

In discussing the nature of the fundamental process which led to the remarkable hypoplasia and deformity of this skeleton, at least four distinct conditions must be considered, namely, rickets, cretinism, chondrodystrophia foetalis, and osteogenesis imperfecta.

The studies of Stilling,¹ Hildebrandt,² and Harbitz³ show very clearly that the disease called by Vrolik and by Stilling osteogenesis imperfecta, which previously in all probability was classed under foetal rickets, is a definite, intrauterine process, the chief characteristics of which are great brittleness and softness of the bones, numerous fractures, and resulting deformities, due to as yet obscure disturbances of myelogenic and periosteal bone formation. The histological studies show that typical lamellar bone tissue and large trabeculae are not formed.

"The trabeculae are small, few in number, far removed from one another, and more or less irregularly arranged; in part they are without mutual connection. A continuous system of trabeculae with lamellar arrangement, Haversian canals, etc., is absent." (Harbitz.) The number of cases is as yet small, and the observations seem to be limited entirely to the bodies of newly-born and very young children. The possibility of recovery from the disease has not been discussed to any extent, and the condition of the skeleton in the event of recovery and continued life cannot be determined from the facts at hand. It is noteworthy, however, that the disease is not necessarily fatal, because Hecker,⁴ Bury,⁵ Klem,⁶ and Railton⁷ have described cases two years, eight months, and one year old, respectively; and Harbitz is inclined to the belief that some of the instances of dwarfism previously referred to foetal rickets, and now regarded as examples of chondrodystrophy, may have been cases of osteogenesis imperfecta. Gurlt⁸ has noted that osteopsathyrosis (fragilitas ossium) occasionally seems to depend upon hereditary conditions, but its possible relation to osteogenesis imperfecta awaits future study.

The condition of the cranium in osteogenesis imperfecta is an interesting one. Vrolik,⁹ J. Schmidt,¹⁰ and Harbitz found that in their cases the calvaria consisted not of continuous bone, but of a mosaic of larger and smaller bone plates, which sometimes touched one another, at other times were united by bridges of periosteum and dura. In Stilling's case the cranium consisted of a membranous sac, with occasional bone spicules here and there. The great number of Wormian bones in the skull of the skeleton now described might well be attributed to such disturbances of cranial ossification. So far as the observations go there seems to be no disturbance at the basal synchondroses in osteogenesis imperfecta.

In the present case the condition of the cranium, the fractures (ribs, humerus), and the evident deficiency of periosteal and myelogenic

¹ Virchow's Archiv, 1899, cxv. pp. 357-370.

² Ibid., 1899, clviii. 426-444.

³ Zeigler's Beiträge, 1901, xxx. pp. 605-628.

⁴ Case abstracted by Harbitz, loc. cit., p. 611.

⁵ Ibid.

⁶ Ibid.

⁷ Ibid.

⁸ Handbuch der Lehre von den Knochenbrüchen, 1862, I. Theil, p. 147.

⁹ Cited by Stilling.

¹⁰ Cited by Harbitz.

ossification in general, favor the notion that it concerns an instance of osteogenesis imperfecta surviving far into adult life. There is, however, relatively only little evidence of previous fractures in the bones of the skeleton. The old fractures in some of the ribs may have been the result of ordinary traumatisms. The number of fractures generally present in the cases described in the literature is usually so large that definite traces would be expected to persist should the patient survive. Virtually all cases of osteogenesis imperfecta so far described have died under one year of age, and it is not impossible that in survivors the traces of healed fractures and infractions eventually may be obliterated or represented by various thickenings.

Harbitz¹ mentions a dissertation by Ekman² that contains a detailed description of a family characterized by the occurrence of dwarfism and osseous fragility through three generations. The ancestor could not walk because of deformity; of his four children, one daughter was small and deformed, with curved arms and legs, and one son was a dwarf. This son had, with a healthy wife, a son that from earliest childhood was liable to fractures on the slightest provocation. He also had curved extremities. This man married a healthy wife and had two children, first a son, who had so many fractures during his youth that when full grown he could not move, and second a daughter, who likewise suffered multiple fractures from early childhood and always remained dwarfed.

While the general appearances of the body in this case may remind one at first glance of a cretin, especially because of the general dwarfing, there is absent the facial expression, the depressed nose, and the thick lips of cretinism, which probably may be ruled out because of the fair degree of intelligence apparently possessed by the individual during life. There is no myxœdema, and the dwarfing is not wholly symmetrical. The total or almost total absence of thyroïdin would favor cretinism, of course, as does the practically complete obliteration of the normal structure of the thyroid.

It is difficult, perhaps wholly impossible, to determine the exact influence which the abnormal state of the thyroid may have exercised upon the man's general health and upon the process of osteogenesis, because of the lack of definite knowledge in regard to his past history and the absence of the evidence that might have resulted from the use of thyroïdin early in life. Perhaps the evil effects of the fibroid transformation of the thyroid were wholly alleviated by compensatory activity on the part of the hypophysis, the weight of which is considerably above the normal average and the structure of which seems fairly normal. The apparently intimate relations between growth of the

¹ Loc. cit.

² *Dissertatio medica descriptionem et casus aliquot osteomalaciæ sistens.* Upsaliæ, 1788.

skeleton and the physiological functions of the thyroid should stimulate to careful chemical and histological study of the thyroid in all instances of general disturbances of skeletal development. In chondrodystrophia foetalis we lack information as to the chemistry of the thyroid, which often has been found abnormal in its appearances. In Harbitz's case of osteogenesis imperfecta (foetus within four to five weeks of full term) the thyroid and thymus did not contain any iodine, but stress cannot be laid on that fact because iodine may be absent normally in the newborn.

The skeleton shows some features that might be explainable on the basis of a healed, severe rickets, such as the beaded ribs, the curvatures, and the flattening of the long bones, the spinal and pelvic deformities. The unusually large number of Wormian bones point to intrauterine disturbances of ossification in the cranial vault. In this case an attempt to trace the condition of the skull to rickets would place the disease at a very early period of intrauterine development, namely, at the sixth to the eighth week, and there seems to be little to warrant such an assumption. The occurrence of congenital rickets, meaning by that rickets which has run its full course before birth, is denied by most observers. As rickets heals the bones become abnormally dense and plump—exactly the reverse of the condition of this skeleton, in which there is also marked enlargements of the articular ends of the long bones. There is absence also of the peculiar deformities (pectus carinatum, funnel-shaped breast) of the thoracic cage, an absence the more significant in view of the profound changes in other parts of the skeleton. True rickety dwarfs usually present long arms, femoral and tibial curvatures. In this case the micromelia is rhizomelic. For these reasons I am inclined to believe that rickets in the ordinary sense cannot explain the case.

Osteomalacia hardly needs to be discussed extensively. As this disease subsides the affected parts may become more or less sclerotic. The shortness of the long bones and the condition of the skull are not explainable on the score of osteomalacia as generally understood.

A glance at the photographs shows that the man was not a true dwarf (microsomia), but a short-limbed or micromelic dwarf. The first general impression probably would be that it concerns an instance of chondrodystrophia foetalis of Kaufmann or achondroplasia (Parrot). This disease, concerning which much has been written of late,¹ was formerly classed as foetal rickets, which term even now is sometimes used expressing the same thing as chondrodystrophia. It is thought to have existed in ancient times, because the statues of the Egyptian god

¹ A fairly good review of the literature is given by Klein in *Centralbl. f. allg. Path. u. Path. Anat.*, 1901, xii. 838-849.

Pthah and other deities are good representations of the external appearances produced by this disease.¹ Chondrodystrophia foetalis affects especially the bones which arise in the cartilage in early foetal life. Indeed, it is said to run its course between the third and sixth weeks, so that it would be stationary or cured when the child is born, while its effect upon the skeleton would remain permanent. The bones especially affected would be, then, those at the base of the skull, the ribs, the pelvis, and the long bones of the extremities. The bones formed entirely in the membrane, and those which, though formed in the cartilage, remain altogether or mainly cartilaginous until a late period of intrauterine life, are usually quite normal in size. Certain changes in the bones of the hands, especially the fingers, described by John Thomson,² William Turner,³ and others, would seem to indicate that the disturbances of ossification may go on at a little later period. The lesions are usually symmetric, the micromelia is rhizomelic; there is some macrocephaly, "trident hands," and normal thorax.

The cause of the disease is wholly unknown. Perhaps B. Morpurgo's⁴ observations on malacic and rickety changes in the skeletons of young white rats, produced by the injection of the cultures of a diplococcus, may prove of value in seeking for explanations of the osseous lesions in rickets, chondrodystrophy, and other diseases. In Morpurgo's animals the lesions at the epiphyseal zones of ossification were similar to those seen in rickets. Regnault⁵ claims that "chien basset" (basset hound) depends upon a disease that corresponds to chondrodystrophia or achondroplasia (Parrot), as the condition is termed in the French literature, in which it also seems to cover osteogenesis imperfecta of the Germans. The peculiar race of dogs referred to has resulted, he says, from the fixation by heredity of an anomaly by means of artificial selection. Leblanc⁶ and Apert⁷ also believe that achondroplasia occurs in animals. "Maitons aneons" (descendants of certain sheep in Massachusetts), "bœufs notos," "veaux-bouledogues," "veaux-tortues," and "chien basset" are the examples given. But in no case has this claim been substantiated by careful histological study. And Cestan points out that here it concerns veritable races capable of transmitting the peculiarities to descendants, whereas achondroplasia in man occurs usually in isolated cases, which rarely transmit the disease, the children, if any are born, being usually normal. History records that the efforts of

¹ Harbitz, loc. cit., note on page 637. Charcot and Richer: Des difformes et les malades dans l'ast, Paris, 1889, p. 12.

² Edinburgh Medical Journal, 1892-93, vol. xxxviii. pp. 1109-1113.

³ Practitioner, 1899, vol. lxxii. pp. 263-277.

⁴ Centralbl. f. Path., 1902, xiii. p. 113.

⁵ Bull. et Mem. de Soc. anat. de Paris, 1901, lxxvi. pp. 386-389.

⁶ Compt. rend. de Soc. de Biol., 1902, liv. 88, 89.

⁷ Ibid., 127-129, and Nouvelle Iconographie de la Salpêtrière, 1901, xiv. 290-298.

Catherine de Medici and others to breed dwarfs did not succeed. Pourak¹ and others, however, have described an instance of apparent transmission of achondroplasia from parent to offspring.

Leblanc² claims that chondrodystrophy often is associated with myxœdema, and that the thyroid probably is the seat of primary disturbance. Nasan³ also ascribes achondroplasia to thyroid dystrophy. This claim is not borne out by the observations of Cestan,⁴ who, in a richly illustrated article, describes a case of chondrodystrophy in a girl, aged nine and one-half years, in which thyroid treatment was given a thorough trial for nine months but without effect, and Marie,⁵ who, in two instances, failed to obtain any benefit from thyroid extract. Christopher⁶ also failed to secure any effect with thyroïodin in one case, and Legny and Regnault⁷ found the thyroid normal in three chondrodystrophic fœtuses. And then there is the fact that the lesions of chondrodystrophy develop in the earliest months of fœtal life before the thyroid is fully developed.

The histological changes which have been studied thoroughly by Kaufmann⁸ and others vary more or less according to the form or variety present, of which Kaufmann recognizes three, namely, the malacic, the hypoplastic, and the hyperplastic. Of the latter only three or four cases are described.⁹ Whether chondrodystrophia malacica, hypoplastic, and hyperplastic are all expressions of the same fundamental disturbance or process may well be doubted. As indicated by the terms used the epiphyseal and other cartilages may be the seat of defective development, of softening, or of overgrowth, the latter being irregular; in all cases normal ossification and growth in length fail to take place, as no columns of cells are formed as normally. In-growth of the periosteum at the lines of ossification may occur, and this may also prevent growth in length.¹⁰ The shafts of the long bones increase in growth from periosteal ossification, which goes on normally in this disease, and the bones become more or less irregular, and, as a rule, thick and plump, hard and sclerotic, as well shown in radiographs, the internal architecture being abnormal. Deformities may arise. Premature synostosis at the base of the skull, and a more or less depressed insertion of the nose, may result.

Two types of physiognomy are spoken of, the cretinoid (premature

¹ Cited from *Nouv. Arch. d'Obst. et de Gyn.*, 1889-90, by Thomson, loc. cit.

² Loc. cit.

³ *Rev. de Neurologie*, 1901, p. 549.

⁴ *Nouvelle Iconographie de la Salpêtrière*, 1901, xiv. 277-289.

⁵ *Presse Medical*, July, 1900, vol. iv.

⁶ *American Medicine*, June 7, 1902.

⁷ *Compt. rend. de Soc. de Biol.*, 1902, liv. 567-568.

⁸ *Sogenannte Fœtale Rickets (Chondrodystrophia Fœtalis)*, Berlin, 1892; Zeigler's *Beiträge*, 1893, xiii. 32-64.

⁹ Kaufmann, Johannesen, Klinger, Joachimsthal.

¹⁰ Collmann. *Virchow's Archiv*, 1901, clxvii. pp. 1-13.

synostosis of the "os tribasillare"), and one marked by flattening of the whole nose rather than retraction of the root of the nose. In Johannesen's case there was no premature basal synostosis.¹ And Regnault also found the cranium normal.² The bones of the calvaria have been found generally normal (Harbitz), or there has been present an exquisite craniotabes (Klein). There may be more or less beading at the costo-chondral junctions. While the trunk is more nearly of the normal length, the extremities are too short, and the skin and soft parts here seem too large. In the hyperplastic form the epiphyseal ends are irregularly swollen. The pelvis is small and deformed, the cotyloid and glenoid cavities may be aplastic, and lordosis seems to be marked; it is explained as due to statical reasons, the weakness of the femurs throwing the weight of the body in front of the normal line. Regnault,³ in a chondrodystrophic fœtus, found the vertebræ involved. Because the infants affected with this strange disease usually die before or shortly after birth (of Kaufmann's thirteen cases only three were born alive), not so very much has been learned as yet concerning the condition of the bones in those that survive. John Thomson⁴ has described two living cases, aged thirty-nine and thirty-six years, the photographs of which in their general features are not unlike the appearances in this case, the noses, however, being much more depressed. The palates were high and arched. There were kyphosis and lumbosacral lordosis, and the limbs were half as long as they should be. The hands had a characteristic spade-like form, being broad and short, the fingers far apart, fusiform and thick, the index and middle fingers turning toward the radial side, the ring and little fingers to the ulnar ("main en trident"). This condition of the hands was absent in this case. Parrot⁵ reports a girl, aged seven and one-half years; William Turner one aged ten years; Boeckh mentions a chondrodystrophic woman whose father and several relatives were similarly affected, and Pourak describes a micromelic female dwarf who became pregnant twice, one of the children showing the same condition as the mother. The disease is probably more frequent than the scanty reports in the literature may be regarded as indicating. The number of cases described has multiplied rapidly of late, especially in French and also in American literature. There is Baldwin's case, to which reference will be made shortly. Osler⁶ describes two cases in brother, aged eleven and a half years, and sister, aged sixteen years. John Lovett Morse reported to the American Pediatric Society at its last meeting (1902)⁷ an in-

¹ Reference cited by Klein, loc. cit.

² Bull et Mem. de Soc. Anat. de Paris, 1901, lxxvi. pp. 419-421, 424-426, and 597-599.

³ Bull et Mem. de la Soc. d'Anat., 1901, lxxvi. pp. 559-560.

⁴ Loc. cit.

⁵ Loc. cit.

⁶ Trans. Congr. Am. Phys. and Surg., 1897, vol. iv. pp. 190-192.

⁷ American Medicine, June 7, 1902.

stance in a child that died at four months. In the discussion Griffith mentioned two cases, Holt one case, and Christopher one case. Abt¹ has described a typical instance in a boy of twelve years, under the title of "Ricketts." This case is interesting, also, because the coccyx was directed forward so as to cause pain on sitting. Cestan,² Apert,³ and others also describe and illustrate living cases. In Cestan's case the micromelia was rhizomelic, the humerus and femurs being most affected. Apert notes that one not seldom sees cases in the streets. Undoubtedly many follow circuses and theatrical troupes. I have seen three apparently typical cases—two men and one woman—on the streets in Chicago, but I was unable to obtain any facts in regard to their history and exact condition. I also know a farmer in Wisconsin who is a micromelic dwarf, four feet and eleven inches in height, and 145 pounds in weight. The fingers of the left hand show the characteristic trident deformity, the fingers on both hands being more nearly the same length than normally and broad. The relative shortness of the limbs is especially marked in the lower extremities. He wears a No. 7½ hat, and the head appears large, the nose being well developed. He is a good worker and presents an athletic appearance, and is a man of intelligence. He was born of healthy parents of usual size; there is no other dwarf in the family. The appearance of chondrodystrophic adolescents and adults are so characteristic that, as Marie⁴ says, if one does not recognize achondroplasia it is because he does not know the disease.

J. P. Baldwin,⁵ in 1890, reported a successful Porro-Cæsarean operation upon a typically rachitic dwarf, weighing 100 pounds, and 47½ inches tall. The illustration accompanying the report shows well the characteristics of chondrodystrophy. The child is stated to have inherited the peculiar deformity of the mother, but no details are given.

Joachimsthal⁶ used the X-ray in the study of dwarfism and allied conditions. In an instance of hyperplastic chondrodystrophy in a girl, aged twelve years, the epiphyses were cartilaginous and swollen, and there were certain peculiar appearances at the ends of the shafts due to the ingrowth of the periosteum, he thought. There was in Joachimsthal's case no growth in length in fifteen months. It is noteworthy that almost without exception those who survived beyond infancy are said to have been of average or more than average intelligence. Many are vigorous physically.

Virchow⁷ has touched upon the conditions here discussed at various

¹ Archives of Pediatrics (reprint).

² Loc. cit.

³ Loc. cit.

⁴ Presse Medicale, July 14, 1900, where he gives an excellent description of chondrodystrophy in adults.

⁵ Medical News, 1890, vol. lvii. pp. 138-141.

⁶ Deut. med. Wochenschr., 1899, xxv. pp. 269-271 and 288-290.

⁷ Virchow's Archiv, 1901, clvi. pp. 192-193; Ibid., 1883, xciv. p. 183.

times. He objects¹ to the term *dystrophia*, introduced by Kaufmann, because the condition imperceptibly shades into a pronounced developmental anomaly (*missbildung*), which, in its most characteristic form, is represented by *phokomelia* (Saint Hilaire, 1836). It concerns rather a profound disturbance in the formation of parts affecting not only cartilage, but also soft parts and bone. (Whether the smallness of the internal organs in cases like the one now reported is due to fundamental genetic disturbances or is secondary to skeletal hypoplasia cannot be determined off hand, and the question merits study.) Congenital rickets leads step by step to *phokomelia*. The name suggested by Kaufmann, and so generally adopted by others, puts into the foreground a single local disease which in turn is referred to a nutritive disorder. He is inclined to think that the name "*foetal rickets*" may yet survive *chondrodystrophy*. As long ago as 1853 Virchow discussed the question of foetal rickets upon the basis of a specimen in the museum at Wurzburg.²

SUMMARY. In the foregoing is described a fairly symmetric micromelic dwarf, aged forty-five years, presumably of fair intelligence; with a fibroid thyroid without any demonstrable thyroiodin; with a relatively large cranial vault made up of 172 Wormian bones, but without any evidence of premature ossification of the synchondroses at the base of the skull; with marked curvatures of the spinal column; with old fractures of many of the ribs and of the left humerus; with great deformity of the pelvis; with curvatures of most of the long bones, which are relatively short, the articular ends being swollen into irregular globular masses; with a general and pronounced osteoporosis; and with absence of trident fingers.

Cretinism or rickets or osteomalacia in all probability is not wholly responsible for the changes present in the skeleton. Taking all the facts and conditions into consideration, it must be acknowledged that the case presents some features best explainable on the score of *osteogenesis imperfecta*, others best explainable on the score of *chondrodystrophia foetalis*, but as for reaching definite conclusion, it is unfortunate that the picture of either of these two diseases, as now understood, is incomplete in some essential particular; and, furthermore, that nothing is known of the man's earlier history. There is absent the premature synostosis of the cranial base of *chondrodystrophia*, and the bones of the extremities are not as plump and hard as would be expected in this disease. While many of the peculiar features correspond well with one's conception of a healed or arrested *osteogenesis imperfecta*, yet there are no observations on record of the bones in survivors from this disease, and there is relatively but little evidence in this case of its

¹ *Zeitschr. f. Ethnologie*, 1898, xxx. p. 55.

² *Virchow's Archiv*, 1853, v. 409-508.

most striking characteristic, namely, extensive multiple fractures. Were a history obtainable of great brittleness and softness of the bones at birth and for some time afterward, the evidence in favor of osteogenesis imperfecta would be materially strengthened. Undoubtedly the porosis of many of the bones in the skeleton may be ascribed to the disuse incident to the deformities, whatever the real nature of the fundamental cause. In this way a more or less atypical chondrodystrophia hyperplastica might have been succeeded by osteoporosis rather than osteosclerosis, as seems to be the rule, but the porosis is so marked and so general as to suggest general genetic disturbances. A combination of osteogenesis imperfecta with chondrodystrophia, complicated later in life by genuine rickets, might also be suggested. As the matter now stands, final conclusion seems unattainable, and it must suffice to place the case upon record as one example of the grave changes that may be produced in the skeleton by obscure and interesting processes, the real cause and nature of which we do not understand.

A NEW METHOD OF CORRECTING FLEXION DEFORMITY AT THE KNEE-JOINT.¹

BY ROYAL WHITMAN, M.D.,
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No deformity is more difficult to correct satisfactorily than fixed flexion at the knee-joint in the class of cases in which one aims to straighten the limb by movement within the joint and to preserve or to increase its range of motion.

In the attitude of flexion the relaxation of the ligaments permits a considerable degree of mobility, a mobility that is increased if the capsule is distended with fluid or if the ligamentous support is weakened or destroyed. Thus flexion deformity is often complicated by backward displacement and outward rotation of the tibia upon the femur. (Fig. 1.)

In the movement from extension to flexion at the knee the articulating surface of the tibia glides backward from the inferior to the posterior portion of the condyles of the femur. If flexion persists, as it practically always does in joint disease, there is subsequent contraction and shortening of the flexor muscles and of the fibrous structures on the posterior aspect of the joint. In many instances there is also an actual enlargement of the lower extremity of the femur, particularly of the inner condyle, and there are other secondary changes

¹ Read at the meeting of the Harvard Medical Society, December 27, 1902.